Arif B Ekici

List of Publications by Year
in descending order

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361 papers	24,920 citations	8755 75 h-index	10734 138 g-index
384 all docs	384 docs citations	384 times ranked	35232 citing authors

#	Article	IF	Citations
1	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
2	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	21.4	960
3	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	13.7	940
4	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
5	CNTNAP2 and NRXN1 Are Mutated in Autosomal-Recessive Pitt-Hopkins-like Mental Retardation and Determine the Level of a Common Synaptic Protein in Drosophila. American Journal of Human Genetics, 2009, 85, 655-666.	6.2	573
6	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	27.0	532
7	Inherited Mutations in 17 Breast Cancer Susceptibility Genes Among a Large Triple-Negative Breast Cancer Cohort Unselected for Family History of Breast Cancer. Journal of Clinical Oncology, 2015, 33, 304-311.	1.6	521
8	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
9	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	6.3	428
10	Genome-wide association studies identify four ER negativeâ€"specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	21.4	374
11	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	12.6	370
12	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
13	Locally renewing resident synovial macrophages provide a protective barrier for the joint. Nature, 2019, 572, 670-675.	27.8	345
14	Common variants at TRAF3IP2 are associated with susceptibility to psoriatic arthritis and psoriasis. Nature Genetics, 2010, 42, 996-999.	21.4	334
15	GWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. Nature Genetics, 2013, 45, 362-370.	21.4	326
16	A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. Nature Genetics, 2010, 42, 874-879.	21.4	321
17	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
18	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptorâ€"negative breast cancer. Nature Genetics, 2011, 43, 1210-1214.	21.4	279

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19	A genome-wide association study identifies a new ovarian cancer susceptibility locus on 9p22.2. Nature Genetics, 2009, 41, 996-1000.	21.4	276
20	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
21	Genome-wide association analysis identifies three new breast cancer susceptibility loci. Nature Genetics, 2012, 44, 312-318.	21.4	256
22	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). Nature Genetics, 2005, 37, 1345-1350.	21.4	252
23	Common variants at 19p13 are associated with susceptibility to ovarian cancer. Nature Genetics, 2010, 42, 880-884.	21.4	235
24	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. American Journal of Human Genetics, 2012, 90, 565-572.	6.2	225
25	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
26	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. American Journal of Human Genetics, 2011, 88, 788-795.	6.2	206
27	Functional Variants at the 11q13 Risk Locus for Breast Cancer Regulate Cyclin D1 Expression through Long-Range Enhancers. American Journal of Human Genetics, 2013, 92, 489-503.	6.2	201
28	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186
29	Genome-wide analyses identify a role for SLC17A4 and AADAT in thyroid hormone regulation. Nature Communications, 2018, 9, 4455.	12.8	181
30	Identification of nine new susceptibility loci for endometrial cancer. Nature Communications, 2018, 9, 3166.	12.8	178
31	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174
32	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. Human Molecular Genetics, 2012, 21, 5373-5384.	2.9	168
33	Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. Human Mutation, 2010, 31, 722-733.	2.5	163
34	CHEK2*1100delC Heterozygosity in Women With Breast Cancer Associated With Early Death, Breast Cancerâ€“Specific Death, and Increased Risk of a Second Breast Cancer. Journal of Clinical Oncology, 2012, 30, 4308-4316.	1.6	162
35	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. Cancer Discovery, 2016, 6, 1052-1067.	9.4	157
36	Low penetrance breast cancer susceptibility loci are associated with specific breast tumor subtypes: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2011, 20, 3289-3303.	2.9	152

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37	Hobit- and Blimp-1-driven CD4+ tissue-resident memory T cells control chronic intestinal inflammation. Nature Immunology, 2019, 20, 288-300.	14.5	152
38	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	6.2	151
39	De Novo Mutations in the Genome Organizer CTCF Cause Intellectual Disability. American Journal of Human Genetics, 2013, 93, 124-131.	6.2	151
40	Inhibiting Interleukin 36 Receptor Signaling Reduces Fibrosis in Mice With Chronic Intestinal Inflammation. Gastroenterology, 2019, 156, 1082-1097.e11.	1.3	148
41	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. Carcinogenesis, 2014, 35, 1012-1019.	2.8	145
42	Epigenetic analysis leads to identification of HNF1B as a subtype-specific susceptibility gene for ovarian cancer. Nature Communications, 2013, 4, 1628.	12.8	144
43	Genetic Regulation of Serum Phytosterol Levels and Risk of Coronary Artery Disease. Circulation: Cardiovascular Genetics, 2010, 3, 331-339.	5.1	141
44	Genome-wide association study identifies a common variant associated with risk of endometrial cancer. Nature Genetics, 2011, 43, 451-454.	21.4	141
45	MicroRNA profiles of prostate carcinoma detected by multiplatform microRNA screening. International Journal of Cancer, 2012, 130, 611-621.	5.1	141
46	Evidence of Geneâ€“Environment Interactions between Common Breast Cancer Susceptibility Loci and Established Environmental Risk Factors. PLoS Genetics, 2013, 9, e1003284.	3.5	136
47	Obstructive airway diseases in women exposed to biomass smoke. Environmental Research, 2005, 99, 93-98.	7.5	132
48	Disease burden and risk profile in referred patients with moderate chronic kidney disease: composition of the German Chronic Kidney Disease (GCKD) cohort. Nephrology Dialysis Transplantation, 2015, 30, 441-451.	0.7	132
49	The German Chronic Kidney Disease (GCKD) study: design and methods. Nephrology Dialysis Transplantation, 2012, 27, 1454-1460.	0.7	127
50	Genome-wide association study identifies a new locus for coronary artery disease on chromosome 10p11.23. European Heart Journal, 2011, 32, 158-168.	2.2	124
51	PU.1 controls fibroblast polarization and tissue fibrosis. Nature, 2019, 566, 344-349.	27.8	121
52	Severely Incapacitating Mutations in Patients with Extreme Short Stature Identify RNA-Processing Endoribonuclease RMRP as an Essential Cell Growth Regulator. American Journal of Human Genetics, 2005, 77, 795-806.	6.2	117
53	Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. BMC Medical Genetics, 2011, 12, 106.	2.1	109
54	Common Breast Cancer Susceptibility Loci Are Associated with Triple-Negative Breast Cancer. Cancer Research, 2011, 71, 6240-6249.	0.9	109

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55	ABCA Transporter Gene Expression and Poor Outcome in Epithelial Ovarian Cancer. Journal of the National Cancer Institute, 2014, 106, .	6.3	107
56	A Peroxisomal Disorder of Severe Intellectual Disability, Epilepsy, and Cataracts Due to Fatty Acyl-CoA Reductase 1 Deficiency. American Journal of Human Genetics, 2014, 95, 602-610.	6.2	106
57	The complement system drives local inflammatory tissue priming by metabolic reprogramming of synovial fibroblasts. Immunity, 2021, 54, 1002-1021.e10.	14.3	106
58	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	12.8	105
59	Molecular differentiation between osteophytic and articular cartilage â€“ clues for a transient and permanent chondrocyte phenotype. Osteoarthritis and Cartilage, 2012, 20, 162-171.	1.3	101
60	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
61	Genetic studies of urinary metabolites illuminate mechanisms of detoxification and excretion in humans. Nature Genetics, 2020, 52, 167-176.	21.4	101
62	19p13.1 Is a Triple-Negativeâ€“Specific Breast Cancer Susceptibility Locus. Cancer Research, 2012, 72, 1795-1803.	0.9	100
63	Risk of Estrogen Receptorâ€“Positive and â€“Negative Breast Cancer and Singleâ€“Nucleotide Polymorphism 2q35-rs13387042. Journal of the National Cancer Institute, 2009, 101, 1012-1018.	6.3	99
64	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	6.3	99
65	Fine-Scale Mapping of the FGFR2 Breast Cancer Risk Locus: Putative Functional Variants Differentially Bind FOXA1 and E2F1. American Journal of Human Genetics, 2013, 93, 1046-1060.	6.2	98
66	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	12.8	98
67	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	5.0	97
68	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 (RSPO4) Cause Autosomal Recessive Anonychia. American Journal of Human Genetics, 2006, 79, 1105-1109.	6.2	94
69	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
70	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	8.2	89
71	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. Kidney International, 2014, 86, 589-599.	5.2	86
72	Prevalence and correlates of gout in a large cohort of patients with chronic kidney disease: the German Chronic Kidney Disease (GCKD) study. Nephrology Dialysis Transplantation, 2015, 30, 613-621.	0.7	85

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73	Homozygosity mapping in 64 Syrian consanguineous families with non-specific intellectual disability reveals 11 novel loci and high heterogeneity. European Journal of Human Genetics, 2011, 19, 1161-1166.	2.8	84
74	Polyol Pathway Links Glucose Metabolism to the Aggressiveness of Cancer Cells. Cancer Research, 2018, 78, 1604-1618.	0.9	83
75	Disturbed Wnt Signalling due to a Mutation in CCDC88C Causes an Autosomal Recessive Non-Syndromic Hydrocephalus with Medial Diverticulum. Molecular Syndromology, 2010, 1, 99-112.	0.8	82
76	Assessing interactions between the associations of common genetic susceptibility variants, reproductive history and body mass index with breast cancer risk in the breast cancer association consortium: a combined case-control study. Breast Cancer Research, 2010, 12, R110.	5.0	82
77	Reduced Syncytin-1 Expression Levels in Placental Syndromes Correlates with Epigenetic Hypermethylation of the ERVW-1 Promoter Region. PLoS ONE, 2013, 8, e56145.	2.5	82
78	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	2.9	80
79	Genome-wide significant risk associations for mucinous ovarian carcinoma. Nature Genetics, 2015, 47, 888-897.	21.4	78
80	Five endometrial cancer risk loci identified through genome-wide association analysis. Nature Genetics, 2016, 48, 667-674.	21.4	77
81	Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376.	2.8	77
82	Fine-Scale Mapping of the 5q11.2 Breast Cancer Locus Reveals at Least Three Independent Risk Variants Regulating MAP3K1. American Journal of Human Genetics, 2015, 96, 5-20.	6.2	76
83	Molecular karyotyping in patients with mental retardation using 100K single-nucleotide polymorphism arrays. Journal of Medical Genetics, 2007, 44, 629-636.	3.2	72
84	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
85	Activation of Epithelial Signal Transducer and Activator of Transcription 1 by Interleukin 28 Controls Mucosal Healing inÂMice With Colitis and Is Increased in Mucosa of Patients WithÂInflammatory Bowel Disease. Gastroenterology, 2017, 153, 123-138.e8.	1.3	72
86	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortiumâ€. Human Molecular Genetics, 2011, 20, 4693-4706.	2.9	71
87	Characterization of germ cell differentiation in the male mouse through single-cell RNA sequencing. Scientific Reports, 2018, 8, 6521.	3.3	70
88	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	3.3	70
89	Identification of the variant Ala335Val of MED25 as responsible for CMT2B2: molecular data, functional studies of the SH3 recognition motif and correlation between wild-type MED25 and PMP22 RNA levels in CMT1A animal models. Neurogenetics, 2009, 10, 275-287.	1.4	68
90	Paradoxical antidepressant effects of alcohol are related to acid sphingomyelinase and its control of sphingolipid homeostasis. Acta Neuropathologica, 2017, 133, 463-483.	7.7	68

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91	Chromatin-Remodeling-Factor ARID1B Represses Wnt/Î²-Catenin Signaling. American Journal of Human Genetics, 2015, 97, 445-456.	6.2	67
92	IL-33-induced metabolic reprogramming controls the differentiation of alternatively activated macrophages and the resolution of inflammation. Immunity, 2021, 54, 2531-2546.e5.	14.3	67
93	Genetic Risk Score Mendelian Randomization Shows that Obesity Measured as Body Mass Index, but not Waist:Hip Ratio, Is Causal for Endometrial Cancer. Cancer Epidemiology Biomarkers and Prevention, 2016, 25, 1503-1510.	2.5	64
94	Variants in RUNX3 Contribute to Susceptibility to Psoriatic Arthritis, Exhibiting Further Common Ground With Ankylosing Spondylitis. Arthritis and Rheumatism, 2013, 65, 1224-1231.	6.7	63
95	Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234.	12.8	63
96	Genetic risk variants for membranous nephropathy: extension of and association with other chronic kidney disease aetiologies. Nephrology Dialysis Transplantation, 2017, 32, 325-332.	0.7	63
97	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
98	Association of Asthma-Related Symptoms With Snoring and Apnea and Effect on Health-Related Quality of Life. Chest, 2005, 128, 3358-3363.	0.8	62
99	CYP19A1 fine-mapping and Mendelian randomization: estradiol is causal for endometrial cancer. Endocrine-Related Cancer, 2016, 23, 77-91.	3.1	62
100	Genetic overlap between endometriosis and endometrial cancer: evidence from crossâ€disease genetic correlation and GWAS metaâ€analyses. Cancer Medicine, 2018, 7, 1978-1987.	2.8	62
101	The 5-HTTLPR polymorphism modulates the influence on environmental stressors on peripartum depression symptoms. Journal of Affective Disorders, 2012, 136, 1192-1197.	4.1	60
102	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	3.5	60
103	Î±-Synuclein-induced myelination deficit defines a novel interventional target for multiple system atrophy. Acta Neuropathologica, 2016, 132, 59-75.	7.7	58
104	Choline transporterâ€like1 (<scp>CHER</scp>1) is crucial for plasmodesmata maturation in Arabidopsis thaliana. Plant Journal, 2017, 89, 394-406.	5.7	58
105	Fatty acid elongation in yeast. Biochemical characteristics of the enzyme system and isolation of elongation-defective mutants. FEBS Journal, 1998, 252, 477-485.	0.2	57
106	A new quantitative PCR multiplex assay for rapid analysis of chromosome 17p11.2-12 duplications and deletions leading to HMSN/HNPP. European Journal of Human Genetics, 2003, 11, 170-178.	2.8	57
107	Five Polymorphisms and Breast Cancer Risk: Results from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 1610-1616.	2.5	57
108	Genome-wide association study with DNA pooling identifies variants at CNTNAP2 associated with pseudoexfoliation syndrome. European Journal of Human Genetics, 2011, 19, 186-193.	2.8	56

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109	MicroRNA profiles classify papillary renal cell carcinoma subtypes. British Journal of Cancer, 2013, 109, 714-722.	6.4	56
110	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	6.3	56
111	BRCA mutations and their influence on pathological complete response and prognosis in a clinical cohort of neoadjuvantly treated breast cancer patients. Breast Cancer Research and Treatment, 2018, 171, 85-94.	2.5	56
112	Breast Cancer Risk â€“ Genes, Environment and Clinics. Geburtshilfe Und Frauenheilkunde, 2011, 71, 1056-1066.	1.8	55
113	ABCB1 (MDR1) polymorphisms and ovarian cancer progression and survival: A comprehensive analysis from the Ovarian Cancer Association Consortium and The Cancer Genome Atlas. Gynecologic Oncology, 2013, 131, 8-14.	1.4	55
114	HOXA10 and HOXA13 sequence variations in human female genital malformations including congenital absence of the uterus and vagina. Gene, 2013, 518, 267-272.	2.2	55
115	Effect of chronic diseases and associated psychological distress on health-related quality of life. Internal Medicine Journal, 2007, 37, 6-11.	0.8	54
116	Common alleles in candidate susceptibility genes associated with risk and development of epithelial ovarian cancer. International Journal of Cancer, 2011, 128, 2063-2074.	5.1	54
117	Inflammation-induced glycolytic switch controls suppressivity of mesenchymal stem cells via STAT1 glycosylation. Leukemia, 2019, 33, 1783-1796.	7.2	54
118	Periodic catatonia: confirmation of linkage to chromosome 15 and further evidence for genetic heterogeneity. Human Genetics, 2002, 111, 323-330.	3.8	53
119	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	2.9	53
120	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	3.3	53
121	Mitochondrial DNA copy number is associated with mortality and infections in a large cohort of patients with chronic kidney disease. Kidney International, 2019, 96, 480-488.	5.2	53
122	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. American Journal of Human Genetics, 2020, 107, 527-538.	6.2	53
123	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
124	Transcription factor Fra-1 targets arginase-1 to enhance macrophage-mediated inflammation in arthritis. Journal of Clinical Investigation, 2019, 129, 2669-2684.	8.2	51
125	Comparison of 6q25 Breast Cancer Hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). PLoS ONE, 2012, 7, e42380.	2.5	51
126	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	6.0	51

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127	Fine-mapping of the HNF1B multicancer locus identifies candidate variants that mediate endometrial cancer risk. Human Molecular Genetics, 2015, 24, 1478-1492.	2.9	50
128	Genotyping NAT2 with only two SNPs (rs1041983 and rs1801280) outperforms the tagging SNP rs1495741 and is equivalent to the conventional 7-SNP NAT2 genotype. Pharmacogenetics and Genomics, 2011, 21, 673-678.	1.5	50
129	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	2.5	49
130	Association Between a Germline OCA2 Polymorphism at Chromosome 15q13.1 and Estrogen Receptorâ€"Negative Breast Cancer Survival. Journal of the National Cancer Institute, 2010, 102, 650-662.	6.3	48
131	Risk of Ovarian Cancer and the NF-Î²B Pathway: Genetic Association with IL1A and TNFSF10. Cancer Research, 2014, 74, 852-861.	0.9	48
132	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	2.7	48
133	Rare Loss-of-Function Mutation in SERPINA3 in Generalized Pustular Psoriasis. Journal of Investigative Dermatology, 2020, 140, 1451-1455.e13.	0.7	48
134	Single-cell RNA sequencing of adult mouse testes. Scientific Data, 2018, 5, 180192.	5.3	48
135	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	7.0	47
136	Mosaicism for the Charcot-Marie-Tooth disease type 1A duplication suggests somatic reversion. Human Genetics, 1996, 98, 22-28.	3.8	46
137	PAX5 biallelic genomic alterations define a novel subgroup of B-cell precursor acute lymphoblastic leukemia. Leukemia, 2019, 33, 1895-1909.	7.2	46
138	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	1.9	45
139	TRIM28 haploinsufficiency predisposes to Wilms tumor. International Journal of Cancer, 2019, 145, 941-951.	5.1	45
140	Combined Associations of a Polygenic Risk Score and Classical Risk Factors With Breast Cancer Risk. Journal of the National Cancer Institute, 2021, 113, 329-337.	6.3	45
141	Common Genetic Variation In Cellular Transport Genes and Epithelial Ovarian Cancer (EOC) Risk. PLoS ONE, 2015, 10, e0128106.	2.5	44
142	FAM13A is associated with non-small cell lung cancer (NSCLC) progression and controls tumor cell proliferation and survival. OncoImmunology, 2017, 6, e1256526.	4.6	44
143	Arginase impedes the resolution of colitis by altering the microbiome and metabolome. Journal of Clinical Investigation, 2020, 130, 5703-5720.	8.2	44
144	Serum levels of miR-320 family members are associated with clinical parameters and diagnosis in prostate cancer patients. Oncotarget, 2018, 9, 10402-10416.	1.8	44

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145	Genetic variants in the tryptophan hydroxylase 2 gene (TPH2) and depression during and after pregnancy. Journal of Psychiatric Research, 2012, 46, 1109-1117.	3.1	43
146	Regulatory eosinophils induce the resolution of experimental arthritis and appear in remission state of human rheumatoid arthritis. Annals of the Rheumatic Diseases, 2021, 80, 451-468.	0.9	43
147	De novo MECP2 duplication in two females with random X-inactivation and moderate mental retardation. European Journal of Human Genetics, 2011, 19, 507-512.	2.8	41
148	Cell-type-specific enrichment of risk-associated regulatory elements at ovarian cancer susceptibility loci. Human Molecular Genetics, 2015, 24, 3595-3607.	2.9	40
149	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	2.9	40
150	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. Scientific Reports, 2018, 8, 4170.	3.3	40
151	EFhd2/Swiprosin-1 is a common genetic determinator for sensation-seeking/low anxiety and alcohol addiction. Molecular Psychiatry, 2018, 23, 1303-1319.	7.9	40
152	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	3.5	39
153	Genome-Wide Association Studies of Metabolites in Patients with CKD Identify Multiple Loci and Illuminate Tubular Transport Mechanisms. Journal of the American Society of Nephrology: JASN, 2018, 29, 1513-1524.	6.1	39
154	Mutations in BRCA1/2 and Other Panel Genes in Patients With Metastatic Breast Cancer â€"Association With Patient and Disease Characteristics and Effect on Prognosis. Journal of Clinical Oncology, 2021, 39, 1619-1630.	1.6	39
155	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. Human Molecular Genetics, 2015, 24, 285-298.	2.9	38
156	Effects of Anti-Integrin Treatment With Vedolizumab on Immune Pathways and Cytokines in Inflammatory Bowel Diseases. Frontiers in Immunology, 2018, 9, 1700.	4.8	38
157	Risk, Prediction and Prevention of Hereditary Breast Cancer â€“ Large-Scale Genomic Studies in Times of Big and Smart Data. Geburtshilfe Und Frauenheilkunde, 2018, 78, 481-492.	1.8	38
158	Rho-A prenylation and signaling link epithelial homeostasis to intestinal inflammation. Journal of Clinical Investigation, 2016, 126, 611-626.	8.2	38
159	Evaluation and impact of chronic cough: comparison of specific vs generic quality-of-life questionnaires. Annals of Allergy, Asthma and Immunology, 2005, 94, 581-585.	1.0	37
160	Polymorphisms in a Putative Enhancer at the 10q21.2 Breast Cancer Risk Locus Regulate NRBF2 Expression. American Journal of Human Genetics, 2015, 97, 22-34.	6.2	37
161	Evidence of a genetic link between endometriosis and ovarian cancer. Fertility and Sterility, 2016, 105, 35-43.e10.	1.0	37
162	11q13 is a susceptibility locus for hormone receptor positive breast cancer. Human Mutation, 2012, 33, 1123-1132.	2.5	35

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163	Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Scientific Reports, 2015, 5, 17369.	3.3	35
164	Mendelian randomization analyses suggest a role for cholesterol in the development of endometrial cancer. International Journal of Cancer, 2021, 148, 307-319.	5.1	35
165	Characterisation of psoriasis susceptibility locus 6 (PSORS6) in patients with early onset psoriasis and evidence for interaction with PSORS1. Journal of Medical Genetics, 2009, 46, 736-744.	3.2	34
166	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	1.2	34
167	Investigation of geneâ€environment interactions between 47 newly identified breast cancer susceptibility loci and environmental risk factors. International Journal of Cancer, 2015, 136, E685-96.	5.1	34
168	Candidate locus analysis of the TERTâ€“CLPTM1L cancer risk region on chromosome 5p15 identifies multiple independent variants associated with endometrial cancer risk. Human Genetics, 2015, 134, 231-245.	3.8	34
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